General

Guideline Title

NSGC practice guideline: prenatal screening and diagnostic testing options for chromosome aneuploidy.

Bibliographic Source(s)

Wilson KL, Czerwinski JL, Hoskovec JM, Noblin SJ, Sullivan CM, Harbison A, Campion MW, Devary K, Devers P, Singletary CN. NSGC practice guideline: prenatal screening and diagnostic testing options for chromosome aneuploidy. J Genet Couns. 2013 Feb;22(1):4-15. [44 references] PubMed

Guideline Status

This is the current release of the guideline.

Recommendations

Major Recommendations

Recommendations for All Patients

- Providers should offer the options of maternal serum screening (MSS) and diagnostic testing for chromosome aneuploidy to every patient.
  - Providers should engage in a discussion with their patients about the benefits, limitations, and risks of MSS and diagnostic testing so that patients may make informed and autonomous decisions.
  - If the provider feels a patient would benefit from additional discussion prior to making a decision, a referral to a genetic counselor or other qualified provider may be appropriate.
  - Documentation of the patient's decision to *elect or to decline* screening and testing should be made in the patient's medical record.
  - Providers should be aware of factors that may impact the options available to their patients, such as the patient's gestational age, insurance coverage, and access to services and providers.
- An ultrasound to assess the fetal anatomy is suggested at approximately 18w0d to 20w0d gestation for all patients regardless of whether or not they choose to have screening or diagnostic testing.

Recommendations for Low Risk Patients Less Than 14 Weeks of Gestation

- For patients who may consider chorionic villus sampling (CVS) or amniocentesis, *stepwise sequential screening or combined first trimester screening* should be considered because:
  - Both are tailored to fit the needs of patients who desire early detection of chromosome aneuploidy but wish to employ a screening method prior to making a decision about diagnostic testing.
  - Both allow for the option of CVS in higher risk pregnancies while deferring testing of lower risk pregnancies to the second trimester

without causing increased anxiety.
    - Of the screening options that provide risk information in the first trimester, stepwise sequential screening has the highest detection rates for Down syndrome and trisomy 18.
- If CVS is not an option, *integrated screening* may be considered in order to maximize detection rates.
- If a patient completes combined first trimester screening, a separate second trimester MSS for chromosome aneuploidy is *NOT* indicated. Screening for chromosome aneuploidy in the second trimester in patients who present prior to 14 weeks should *ONLY* be performed as a part of integrated, serum integrated, stepwise sequential, or contingency screening. Independent screening in first and second trimesters increases the false positive rate of screening.
- Patients who have an increased nuchal translucency (NT) (≥95th% or ≥3.0 mm) should be offered diagnostic testing by either CVS or amniocentesis. A referral for a fetal echocardiogram should also be considered if the NT ≥3.5 mm.
- Early amniocentesis (prior to 15 weeks of gestation) is *not* recommended due to the increased risks for pregnancy loss, clubfoot, and fluid leakage. CVS should be offered as the diagnostic testing option for chromosome aneuploidy in the first trimester.

Recommendations for Low Risk Patients After 14 Weeks of Gestation

- Patients who desire MSS but did not have MSS in the first trimester should be offered a *quad* or *penta* screen rather than a triple screen due to the increased detection rates.
- Amniocentesis should be offered as the diagnostic testing option for chromosome aneuploidy for patients after 15 weeks of gestation.

Recommendations for Patients at Increased Risk for Chromosome Aneuploidy

- Patients who desire screening information may be offered non-invasive prenatal testing (NIPT) due to the high detection rates and low false positive rates. NIPT should only be offered in the context of informed consent, education, and counseling by a qualified provider, such as a genetic counselor. Standard confirmatory diagnostic testing should be offered as follow-up to positive NIPT results. High risk patients who decline NIPT but remain interested in screening should be made aware of alternate screening options as appropriate based on gestational age and screening availability.
- If the patient presents prior to 14 weeks gestation, CVS and amniocentesis should both be offered as diagnostic testing options for chromosome aneuploidy.
- If the patient presents after 14 weeks gestation, amniocentesis should be offered as the diagnostic testing option for chromosome aneuploidy.

Clinical Algorithm(s)

A decision tree for selecting between screening and diagnostic testing options is provided in the original guideline document.

Scope

Disease/Condition(s)

Chromosome aneuploidy

Guideline Category

Counseling

Diagnosis

Risk Assessment

Screening

Clinical Specialty

Family Practice

Medical Genetics

Obstetrics and Gynecology

Intended Users

Advanced Practice Nurses

Allied Health Personnel

Health Care Providers

Physician Assistants

Physicians

Psychologists/Non-physician Behavioral Health Clinicians

Social Workers

Guideline Objective(s)

To provide information that assists physicians and allied health professionals in making decisions about different screening and diagnostic testing for chromosome aneuploidy throughout pregnancy

Target Population

Pregnant patients who are in the first or second trimester

Interventions and Practices Considered

1. Discussion of benefits, limitations, and risks of maternal serum screening
2. Referral to genetic counselor
3. Documentation of patient's decision to elect or refuse screening
4. Ultrasound to assess fetal anatomy
5. Chorionic villus sampling (CVS)
6. Amniocentesis (early amniocentesis is not recommended)
7. Noninvasive prenatal testing

Major Outcomes Considered

Not stated

Methodology

Methods Used to Collect/Select the Evidence

Searches of Electronic Databases

Description of Methods Used to Collect/Select the Evidence

The guideline authors searched via PubMed for articles from 2008 to 2012, but articles from any year were used, including articles from the 1970s and 1980s. Articles on prenatal chromosome testing and screening were used as well as a mix of landmark papers, original research, and reviews in the publication. Specific terms included (but were not limited to): prenatal diagnosis, prenatal screening, chromosome aneuploidy, first trimester screening, integrated screening, contingency screening, stepwise sequential screening, maternal serum screening, ultrasound for chromosome aneuploidy, CVS, amniocentesis, NIPT, NIPD, cell free fetal DNA, and genetic counseling for prenatal testing.

Number of Source Documents

79

Methods Used to Assess the Quality and Strength of the Evidence

Expert Consensus (Committee)

Rating Scheme for the Strength of the Evidence

Not applicable

Methods Used to Analyze the Evidence

Review

Description of the Methods Used to Analyze the Evidence

Not stated

Methods Used to Formulate the Recommendations

Expert Consensus

Description of Methods Used to Formulate the Recommendations

Not stated

Rating Scheme for the Strength of the Recommendations

Not applicable

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Internal Peer Review

Description of Method of Guideline Validation

Not stated

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of evidence supporting the recommendations is not specifically stated.

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

- Appropriate use of prenatal screening and diagnostic testing options for chromosome aneuploidy
- The primary benefit of chorionic villus sampling (CVS) is that it can be performed at an earlier gestational age, allowing for earlier decision-making.

Potential Harms

- The primary limitation of screening is that it does not provide a definitive diagnosis, leading to the potential of increased anxiety in women with an unaffected pregnancy and the potential of false reassurance in women who have a pregnancy with a chromosome aneuploidy. Another limitation of screening is the variability in the detection rates, false positive rates, screening cut-offs, and anatomical ultrasound markers included in the screen based on the particular laboratory and/or provider involved. In addition, detection rates for screening in multiple gestations are generally decreased from those of singletons.
- Despite numerous potential benefits associated with screening and testing for chromosome aneuploidy, there are also limitations. Available options may be limited by a number of factors, including: gestational age of the patient at entry into the healthcare system, state regulations impacting available options, insurance coverage and out-of-pocket costs to the patient, laboratory contracts, availability of laboratory draw sites, access to certified nuchal translucency (NT) providers, and access to physicians who perform chorionic villus sampling (CVS) or amniocentesis. One of the main barriers faced in screening is insurance coverage, as each private and public insurance plan has specific requirements for coverage of screening and diagnostic testing for chromosome aneuploidy. Additionally, there is often a time lapse between the publication of guidelines recommending the incorporation of a new test and routine coverage of that test by insurance companies. These issues should be considered when assessing a patient's access to various prenatal screening and diagnostic testing options.

Contraindications

Contraindications

Early amniocentesis (prior to 15 weeks of gestation) is *not* recommended due to the increased risks for pregnancy loss, clubfoot, and fluid leakage.

Qualifying Statements

Qualifying Statements

- The practice guidelines of the National Society of Genetic Counselors (NSGC) are developed by members of the NSGC to assist genetic counselors and other health care providers in making decisions about appropriate management of genetic concerns, including access to

and/or delivery of services. Each practice guideline focuses on a clinical or practice based issue, and is the result of a review and analysis of current professional literature believed to be reliable. As such, information and recommendations within the NSGC practice guidelines reflect the current scientific and clinical knowledge at the time of publication, are only current as of their publication date, and subject to change without notice as advances emerge.

- In addition, variations in practice, which take into account the needs of the individual patient and the resources and limitations unique to the institution or type of practice, may warrant approaches, treatments, and/or procedures that differ from the recommendations outlined in this guideline. Therefore, these recommendations should not be construed as dictating an exclusive course of management, nor does the use of such recommendations guarantee a particular outcome. Genetic counseling practice guidelines are never intended to displace a health care provider's best medical judgment based on the clinical circumstances of a particular patient or patient population. Practice guidelines are published by NSGC for education and informational purposes only, and NSGC does not "approve" or "endorse" any specific methods, practices, or sources of information.

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Clinical Algorithm

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

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Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2013 Feb

Guideline Developer(s)

National Society of Genetic Counselors - Medical Specialty Society

Source(s) of Funding

National Society of Genetic Counselors

Guideline Committee

National Society of Genetic Counselors Practice Guidelines Committee

Composition of Group That Authored the Guideline

Authors: K. L. Wilson; J. L. Czerwinski; J. M. Hoskovec; S. J. Noblin; C. M. Sullivan; A. Harbison; M. W. Campion; K. Devary; P. Devers; C. N. Singletary

Financial Disclosures/Conflicts of Interest

Not stated

Guideline Status

This is the current release of the guideline.

Guideline Availability

Electronic copies: Available to subscribers from the Journal of Genetic Counseling Web site .

Availability of Companion Documents

The following is available:

- Bennett RL, French KS, Resta RG, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. J Genet Couns 2008 Oct;17(5):424-33. Available to subscribers from the Journal of Genetic Counseling Web site .

Patient Resources

None available

NGC Status

This NGC summary was completed by ECRI Institute on May 28, 2013. The information was verified by the guideline developer on June 18, 2013.

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